Allan Award Introduction: Arthur L. Beaudet

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It is truly a pleasure and privilege to introduce this year's recipient of the William Allan Award, Arthur L. Beaudet, who is the Henry and Emma Meyer Professor and Chairman of the Department of Molecular and Human Genetics at the Baylor College of Medicine, in Houston, Texas. Art earned his first degree from the College of the Holy Cross and his M.D. from Yale University, graduating from both with honors. After internship and residency on the Harriet Lane Pediatric Service at the Johns Hopkins Hospital, he went to the National Institutes of Health to do research at a time when there was tremendous political turmoil in our country with the Vietnam War. His lab mates and colleagues at the NIH included Joe Goldstein (a former recipient of this award), Tom Caskey, Ed Skolnick, and Marshall Nirenberg; Art worked on very basic mechanisms of protein translation. In 1971, he came to the Baylor College of Medicine, where he has spent almost 37 years contributing to human and medical genetics and the last 13 years as our leader as Chair of the Department.

I first met Art 22 years ago when I came to Baylor to interview for my Pediatric residency and clinical genetics fellowship. In my greater than two decades there, Art has been an inspiration, a great mentor, and a terrific leader. However, we his colleagues, collaborators, and friends in the Society for whom he was a former president recognize him today for his scientific contributions.

William Allan was a pioneering human/medical geneticist whose primary "high-tech" experimental approach for research was pedigree analysis. I thought this would be fun to mention since all of us who did our clinical training with Art were drilled by Art until we got the pedigree correct. I made this slide from one of William Allan's papers[1], where he posited two rules from such pedigree studies. (1) Regarding diseases due to a single gene defect, a survey of 50–100 families usually shows a triple pattern of inheritance: dominant, recessive, and X-linked. I think this is perhaps the first recognition of genetic heterogeneity. (2) The pattern of inheritance determines the age of onset and clinical severity as demonstrated by Charcot-Marie-Tooth disease (CMT). Allan made many important observations on CMT and retinitis pigmentosa.

The Allan Award is given by our Society "to recognize substantial and far-reaching scientific contributions to human genetics, carried out over a sustained period of scientific inquiry and productivity". Art Beaudet began his research career with fundamental studies on protein synthesis.[2,3] During the next four decades, among many other research accomplishments, he delineated the molecular genetics of inborn errors of metabolism, focusing on urea cycle defects, for which he has also worked on gene therapy.[4–6]

Art also described the first case of uniparental disomy (UPD) as a genetic mechanism for disease, which was published in our Society's Journal.[7] The full contribution of the UPD mechanism to disease may become even more realized as we see better array-based genomic assays applied in the clinic. Art has actively pursued imprinting in Prader-Willi and Angelman syndromes[8–10] and more recently in autism as one aspect of his mixed epigenetic, genetic, de novo and inherited, or so-called MEGDI, model.[11,12] I imagine that William Allan would have loved this model to explain genetic heterogeneity in autism. Finally, Art spearheaded the clinical implementation of high-resolution genome analysis by array comparative genomic hybridization.[13–15]

His sustained period of scientific inquiry and productivity spans four decades and includes over 300 publications and the training of about 70 postdoctoral fellows and 10 graduate students in his own laboratory. Moreover, as director for more than 30 years of the largest clinical genetics training program, he has overseen the training of greater than 120 clinical genetics fellows from the United States and throughout the world including fellows from over 25 countries.

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I want to personally thank the Society for the wisdom of its choice. William Allan would have been proud of Art's pedigree analysis. I know we will all enjoy hearing Art's perspective.

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